

TABLE 1
Genetic variants and their associated clinical significance in the context of the human genome

SEQ ID	Sequence Calling Assembly	Base pos. of SNP	Polymorphic Sequence	Base before	Base after	Amino Acid before	Amino Acid after	Type of Change	Protein classification of CuraGen gene	Name of protein identified following a BLASTX analysis of the CuraGen sequence	p value	Allele Freq. (pred.)	Map Location	Therapeutic Area #1	Therapeutic Area #2
1, 2	cg44928667	1090	TGGAGCAGAAAGG TGGAGCTGGACT CCAGG[<i>gap</i>]/TATC TGAGTG[CTGCGC TGAAGAAATACC AGACTGAGCAAA GGAG	gap (1)	TATCT GAGT G(2)	Arg	Lys	Frameshift	kinase receptor	Human Gene SPTREMBL-ID Q60437 INSULIN RECEPTOR TYROSINE KINASE 53 KDA SUBSTRATE - UNKNOWN, 521 aa	4.60E-246	deletion of 10bp in 3 of 11	17	Metabolic/endocrine/cardiovascular	
3, 4	cg43957213	1529	CATACATAAACG GGCAAGATTTCAG TCCCTGACCGCA A[<i>gap</i>]/GJGCA- CTTACAGTCTAG TTGGGAAGGGAG ACACAAAT	gap (3)	G (4)			Silent, Non-Coding	misc_channel	Human Gene SWISSNEW-ID P37088 AMILORIDE-SENSITIVE SODIUM CHANNEL ALPHA-SUBUNIT (LUNG NA+ CHANNEL ALPHA SUBUNIT) (ALPHA ENAC) (NONVOLTAGE-GATED SODIUM CHANNEL 1 ALPHA SUBUNIT) (SCNEA) (ALPHA NACH) - HOMO SAPIENS (HUMAN), 669 aa [pels SWISSPROT-ID P37088 AMILORIDE-SENSITIVE SODIUM CHANNEL ALPHA-SUBUNIT (LUNG NA+ CHANNEL ALPHA SUBUNIT) (ALPHA ENAC) (NONVOLTAGE-GATED SODIUM CHANNEL 1 ALPHA SUBUNIT) (SCNEA) (ALPHA NACH) - HOMO SAPIENS (HUMAN), 669 aa [pels TREMBLNEW-ID E308262 AMILORIDE-SENSITIVE EPITHELIAL SODIUM CHANNEL ALPHA SUBUNIT - HOMO SAPIENS (HUMAN), 669 aa		good 10 of 20	1p36.1	Metabolic/endocrine/cardiovascular	Renal Disease
5, 6	cg44912878	1164	ACCGCATCATGG AGGTCATCGATG[T/C]/CATCACCAC CACTGCCCAGAG CCACC	T (5)	C (6)	Ala	Val	CONSERVATIVE	kinase	Human Gene SWISSPROT-ID P17858 6-PHOSPHOFRUCTOKINASE, LIVER TYPE (EC 2.7.1.11) (PHOSPHOFRUCTOKINASE 1) (PHOSPHOHEXOKINASE) (PHOSPHOFRUCTO-1-KINASE ISOZYME B) - HOMO SAPIENS (HUMAN), 780 aa		good 2 of 9	21	Metabolic/endocrine/cardiovascular	

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7, 8	cg44912878	3171	CT-A- TCCCTGGCCACC TG-CCAGGCTCC CTC[G/C]GGCTGG TGCTT-GAGA- CCA-GCCTG- CCAGGCC	G (7)	C (8)			3' UTR	kinase	Human Gene SWISSPROT-ID P178586 PHOSPHOFRUCTOKINASE, LIVER TYPE (EC 2 7 1 1) (PHOSPHOFRUCTOKINASE 1) (PHOSPHOHEXOKINASE) (PHOSPHOFRUCTO-1-KINASE ISOZYME B) - HOMO SAPIENS (HUMAN), 780 aa		good 7 of 20	21	Metabolic/endocrine/cardiovascular	
9, 10	cg44912878	853	ATCATCCAGCTG GGCGGCACTATC ATTGGCAGCGCT CGCT[CG/GC]AAG GCCTTACCACC AGGGAGGGGCG CCGGGCAGCGG	CG (9)	GC (10)	Cys	Ser	CONSERVATIVE	kinase	Human Gene SWISSPROT-ID P178586 PHOSPHOFRUCTOKINASE, LIVER TYPE (EC 2 7 1 1) (PHOSPHOFRUCTOKINASE 1) (PHOSPHOHEXOKINASE) (PHOSPHOFRUCTO-1-KINASE ISOZYME B) - HOMO SAPIENS (HUMAN), 780 aa	1.330E-06	good 4 of 8	21	Metabolic/endocrine/cardiovascular	
11, 12	cg44921974	304	TTGAGTTCGGTC ACAGACTTGATG TTTTT-GA- AAG[C/T]TGTAC CAGTTTATTGTCA CCTTCCAAGTGA ACCACTGCTTG	C (11)	T (12)	Thr	Ala	CONSERVATIVE	UNCLASSIFIED	Human Gene Similar to SWISSPROT-ACC P07148 FATTY ACID-BINDING PROTEIN, LIVER (L-FABP) - Homo sapiens (Human), 127 aa	1.130E-06	9 of 30	2 (4q28)	Metabolic/endocrine/cardiovascular	
13, 14	95124747	643	aaagtgggttcagagctt ctttccctaattg[c/t]ggg cctcaccattgcatgcaatg actatttttagtacatgaa gcagaagggaagaagla gg	C (13)	T (14)	Ala	Val	CONSERVATIVE		Calpactin	1.170E-11	44 of 1050	17	Metabolic/endocrine/cardiovascular	

TABLE 1
Genetic variants associated with type 2 diabetes mellitus

SEQ ID	Sequence Calling Assembly	Base pos. of SNP	Polymorphic Sequence	Base before	Base after	Amino Acid before	Amino Acid after	Type of Change	Protein classification of CuraGen gene	Name of protein identified following a BLASTX analysis of the CuraGen sequence	p value	Allele Freq. (pred.)	Map Location	Therapeutic Area #1	Therapeutic Area #2
15, 16	88073933	751	GGAGTGGGGCTA CGCCAGCCACAA CGGTCTCTGACCA CTGGCATGAACT TTTCCCAAATGC CAAGGGGGAAA ACCAAGTCGCC[A] /G]TTGAGCTGCA TACTAAAGACAT CAGGCATGACCC TTCTCTGCAGCC ATGGTC- TGTGTCTTATGAT GGTGG	A (15)	G (16)	Ile	Val	CONSERVATIVE		Carbonic Anhydrase 3		5 of 12	8	Metabolic/endocrine/cardiovascular	
17, 18	cg43953338	1246	AATTTGGG-TGGT TTGAAAGGATCAC ATAAA- GGAGATCCAGA[A/G]A-TGCC- GGCGTTTGATTCT TATTGCTT	A (17)	G (18)			Silent-Coding	synthase	Human Gene Homologous to SWISSPROT-ID P44708 GLUCOSAMINE--FRUCTOSE-6-PHOSPHATE AMINOTRANSFERASE (ISOMERIZING) (EC 2 6 1 16) (HEXOSEPHOSPHATE AMINOTRANSFERASE) (D-FRUCTOSE-6- PHOSPHATE AMIDOTRANSFERASE) (GFAT) (L-GLUTAMINE-D-FRUCTOSE-6-PHOSPHATE AMIDOTRANSFERASE) (GLUCOSAMINE-6-PHOSPHATE SYNTHASE) - HAEMOPHILUS INFLUENZAE, 609 aa	3 10E-107	4 of 40	2 (2p13)	Metabolic/endocrine/cardiovascular	
19, 20	cg43953338	3084	ATGTTGGGTATC CTAC- TACTTTGTGTTTT CATCTCCTAAAA GTG[G/TTTTTA TTTCCTTGTAICT GTAGTCITTTATT TTTTAAATGAC	G (19)	T (20)			3' UTR	synthase	Human Gene Homologous to SWISSPROT-ID P44708 GLUCOSAMINE--FRUCTOSE-6-PHOSPHATE AMINOTRANSFERASE (ISOMERIZING) (EC 2 6 1 16) (HEXOSEPHOSPHATE AMINOTRANSFERASE) (D-FRUCTOSE-6- PHOSPHATE AMIDOTRANSFERASE) (GFAT) (L-GLUTAMINE-D-FRUCTOSE-6-PHOSPHATE AMIDOTRANSFERASE) (GLUCOSAMINE-6-PHOSPHATE SYNTHASE) - HAEMOPHILUS INFLUENZAE, 609 aa	3 10E-107	4 of 10	2 (2p13)	Metabolic/endocrine/cardiovascular	

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SEQ ID	Sequence Calling Assembly	Base pos. of SNP	Polymorphic Sequence	Base before	Base after	Amino Acid before	Amino Acid after	Type of Change	Protein classification of CuraGen gene	Name of protein identified following a BLASTX analysis of the CuraGen sequence	p value	Allele Freq. (pred.)	Map Location	Therapeutic Area #1	Therapeutic Area #2
21, 22	cg42930646	1229	TGTCAGCCCCAC AAATAGGAGTGG TCAATGTTACTG ATGCGGATAGCG TA- TGGATGGAA[A/G JTGGACGATGAG GAGGACCTGCCT TCTGCTGAGGAG CTGGAGGACTGG CTGGAGG	A (21)	G (22)	Met	Val	CONSERV ACTIVE	lamunin	Human Gene SWISSPROT-ID P07221 CALSEQUESTRIN, SKELETAL MUSCLE ISOFORM PRECURSOR (ASPARTACTIN) (LAMININ- BINDING PROTEIN) - ORYCTOLAGUS CUNICULUS (RABBIT), 395 aa	1.80E-198	4 of 19	1	Metabolic/endocrine/ cardiovascular	Bone Disease
23, 24	94842816	1303	CCGCAGTTCCT CTTCCCACGACT CAGAGCCCACCTT[A/T]TCCACTTCT TTCGAAACTCCG ACGCGACATCCA ACCGAGCGGTGT CAGC	A (23)	T (24)	Asn	Lys	NON- CONSERV ACTIVE	Secreted hormone	Adrenomedullin		4 of 55	11	Metabolic/endocrine/ cardiovascular	
25, 26	88048627	3294	CCTATTACCAGA GAGGATCGAGCA TGGTCCTCTTCTC C[C/T]C[C]CCACCT GTGATCCTTCCTG ATCTCTTCTCA TCCTCCTGATAGT GGGATGA	C (25)	T (26)	Pro	Ser	NON- CONSERV ACTIVE	Membrane protein	Prion protein (new variant)		2 of 16	20p12.1-13	Metabolic/endocrine/ cardiovascular	CNS Disorders
27, 28	88048627	2968	GGTGCTGCAGCA GCTGGGGCAGTG GTGGGGGGCCTT GGCGGCTA- C[A/G]TGCTGGGA AGTGCCATGAGC AGGCCCATCATA CAAT	A (27)	G (28)			Silent- Coding	Membrane protein	Prion Protein (previously identified variant)		3 of 9	20p12.1-13	Metabolic/endocrine/ cardiovascular	CNS Disorders
29, 30	95124747	304	Atgccatcctaaatgaac acgccatggaaccatgat gtttacatttcaca[a/c]attc gctggggataaaggctactt aacaaaggaggacctgaga gtactcatggaaggag	A (29)	C (30)	Lys	Glu	NON- CONSERV ACTIVE		Calpactin		184 of 350	17	Metabolic/endocrine/ cardiovascular	

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31, 32	95124747	331	Atgccatctcaaatggaac acgcacatggaacacatgat gtttacattcacaaattcgct ggggataaag[g/a]ctactt aacaanaggaggacctgaga gtactcatggaaaaggag	G (31)	A (32)	Gly	Asp	NON- CONSERV ATIVE		Calpactin		8 of 900	17	Metabolic/endocrine/cardiovascular	
33, 34	91234048	300	gccccaggatgggtgagtt caacgagaagaagacaac atgtggcaccgtt[g/a]cct caagtaccctgctgttaccta caattgctgcttcctggcgtggc tgg	G (33)	A (34)	Cys	Tyr	NON- CONSERV ATIVE		CD151	2.20E-11	3 of 260		Metabolic/endocrine/cardiovascular	Immunology
35, 36	91234048	965	tgtagaccagcgtgtggacc agctgcagcaggagttcc[a /c]ctgctgtggcagcaaca actcacaggactggcgaga cagtagagtggtatccg	A (35)	C (36)	His	Pro	NON- CONSERV ATIVE		CD151	3.00E-10	6 of 200		Metabolic/endocrine/cardiovascular	Immunology
37, 38	94218949	1446	TAC-G-AGAAGG- CGAC-G-ATG-A- CCGG- ACTGTGTGCCCGG G-AG-A-T- CCG[C/T]CACAA- CT-CCACGGG- CTGCC-T-GCGGA- T-G-AA-GG-ACCA- GTG-T-G-AC	C (37)	T (38)			Silent- Coding		Clustern/ApoJ	8.50E-10	6 of 100	8p12-21	Metabolic/endocrine/cardiovascular	

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39, 40	95351416	1876	GTCAA-TGTCGG-TTTAC-TG-TACACCA-AA-TAAA-CCA-AGCAGG-AC-ATCAA-TGAGC-AA---G[A/G]T-GTTTGTGAA-GGG-TG-CTCC-TG-AA-GG-TG-TCA-TTGACA-GG-T-GCACCC-A-CA-TTCGA-G-TT	A (39)	G (40)	Met	Val	NON-CONSERVATIVE		SerCA1	8.00E-14	3 of 130	12q24.1	Metabolic/endocrine/cardiovascular	
41, 42	97873686	1132	gaagacattgttggtgcaggaagcagctatg/tctcagataaaggagatgttggaactgccccctgagacatccgccccttaaggccaattgg	G (41)	T (42)	Ala	Ser	NON-CONSERVATIVE		Valosin-containing protein	5.60E-08	5 of 600	1	Metabolic/endocrine/cardiovascular	
43, 44	95292679	1232	AA-TG-ATAAC-TTC-TTTTG--A-GGGG-AA-GG-A-GCTG-C-GG-C-TG-AAGC-A-GG-AGT-AC TT-C-G-T-GGTGG-CC[G/T]CCA-CGCT-C-C-AGGAC-AT-C-AT-CCGC-C-G-C--TTCAAGT-C-G-T-CCAAG-TTCGG-CTG-CCGGGA-CCC-TG	G (43)	T (44)	Ala	Ser	NON-CONSERVATIVE		Glycogen Phosphorylase Muscle	2.10E-14	24 of 120	20	Metabolic/endocrine/cardiovascular	

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45, 46	91231553	1353	TGATGGATAAATT CCCGGAATGCTC CTTTGGCTGGTTT TGGTACGGCTT GCCAATTTCCTCGT CTGTATGC[A/C]A AGTAC-TTTCAA- GGAGATCT-GAAT- CTC-TACTC- TTTAT-C-AGGA- TATGG-AACAGA- TGCTAT	A (45)	C (46)			Silent-Coding		Pyruvate dehydrogenase kinase-like protein	6.50E-17	6 of 10	7q21-q22	Metabolic/endocrine/cardiovascular	
47, 48	95108682	2804	CT- GGACCTGATTT- CC- TGACCACAGGC- TCTTGAAAG[T/C]- CCCCATGGT- CTTGCTGAC-AGA- GG- CCCCTAGAGTAA AAGGAGC	T (47)	C (48)			SILENT NONCODING		Galactosidase sialotransferase		7 of 45	3q27	Metabolic/endocrine/cardiovascular	
49, 50	91234048	227	ggggagcttctgtccactg tcctgcagaggagtcgttcc cagcccggc[t/a]gcccca ggatgggtgagttcaacga gaagaagacaacatgtggc accgttt	T (49)	A (50)			Silent-Coding		CD151	0.23	91 of 250		Metabolic/endocrine/cardiovascular	Immunology
51, 52	94131544	84	ATTAAAGATTTG ATTATTCAAGTA TGTGAAAACATT CTACAATGGAAA CT[C/G]TTATTAG ATGCTGCATGTA CTGTGCTATGGA- CCAC-GCACAT- ACAGCC- ATGCTGTTTC- AGAAGAC	C (51)	G (52)			SILENT NONCODING		Rab5-interacting protein		5 of 50	20	Metabolic/endocrine/cardiovascular	

TABLE 1
Genetic variants in the CYP2C9 gene and their association with warfarin sensitivity

SEQ ID	Sequence Calling Assembly	Base pos. of SNP	Polymorphic Sequence	Base before	Base after	Amino Acid before	Amino Acid after	Type of Change	Protein classification of CuraGen gene	Name of protein identified following a BLASTX analysis of the CuraGen sequence	p value	Allele Freq. (pred.)	Map Location	Therapeutic Area #1	Therapeutic Area #2
53, 54	95343665	909	CACCTC-CCT- CACCACACAGGA CCCTGAGT- GAGGA- GGAGGGGCTGGA AACCTGGG[G/C]T GGGTGGCCAAA GGAGAACCTCAG GCTCCTGGCCTG GCCAGCTCCTT CCTGCCCAAGGT AGCTTAGCCCAT CC	G (53)	C (54)			SILENT NONCODI NG		Adipocyte-specific protein		3 of 12	3p	Metabolic/endocri ne/cardiovascular	
55, 56	97873686	3429	CACAGCCCTGCTC CATTC1CCAG- TCTGAACAGTTC AGCTA- CAGTCTGACTCT GGACA- GGG[G/T]GTTT- CTGTTC- AAAAATACAAAA CAAAAGCGATAA AATAAAG- CGATTTCATT	G (55)	T (56)			SILENT NONCODI NG		Valosn-containing protein		9 of 30	9p13	Metabolic/endocri ne/cardiovascular	Renal Disease
57, 58	97978029	2183	AGCTTGCCTTAA ATTATTTTATAT GACTGTGTGCTCT CTAGGTAGCC- TTTGGTCTATTGT ACACAA[T/C]CTC ATTTCATATGTTT GCATTTTGGCAA AGAACTTAATAA AATTGTTCAGTG	T (57)	C (58)			SILENT NONCODI NG		Medium Chan Acyl Coa Dehydrogenase		4 of 11	1p31 1-31 3	Metabolic/endocri ne/cardiovascular	
59, 60	95289295	3121	aaggccaccatgcttttatta tcgcttig[c/gap]lggaga caaaagcacaagctccgagt gtgctgggagctctccatta actagag	C (59)	gap (60)			SILENT NONCODI NG		Creatine Kinase Muscle		25 of 90	19q13 2	Metabolic/endocri ne/cardiovascular	

TABLE 1. Genomic and clinical characteristics of the four patients with rare forms of congenital hypothyroidism.

SEQ ID	Sequence Calling Assembly	Base pos. of SNP	Polymorphic Sequence	Base before	Base after	Amino Acid before	Amino Acid after	Type of Change	Protein classification of CuraGen gene	Name of protein identified following a BLASTX analysis of the CuraGen sequence	p value	Allele Freq. (pred.)	Map Location	Therapeutic Area #1	Therapeutic Area #2
61, 62	cg42709360	955	GGCCGGGGAGTG GCGATGGTGACT GC[C/T]GTGGCTG CCCGTCTGGCTG CCCACCG	C (61)	T (62)			Silent-Coding	kinase	Human Gene Similar to SWISSPROT-ID P17709 GLUCOKINASE (EC 2.7.1.2) (GLUCOSE KINASE) (GLK) - SACCCHAROMYCES CEREVISIAE (BAKER'S YEAST), 500 aa pels SWISSPROT-ID P17709 GLUCOKINASE (EC 2.7.1.2) (GLUCOSE KINASE) (GLK) - SACCCHAROMYCES CEREVISIAE (BAKER'S YEAST), 500 aa	4.10E-39		5 (5q35.2)	Metabolic/endocrine/cardiovascular	
63, 64	cg43920091	21	GGCGCCTAGGTT GTGTTGAGAGGG GGATGCCCTG[G /gap]CCCTGCCTC ACTGTGACCTG- CTCCTGCCCCACG- TGC	G (63)	gap (64)			SILENT-NONCODING	ATPase-associated	Human Gene SWISSPROT-ID Q13608 PEROXISOME ASSEMBLY FACTOR-2 (PAF-2) (PEROXISOMAL-TYPE ATPASE 1) (PEROXIN-6) - HOMO SAPIENS (HUMAN), 980 aa	0		6 (6p21.1)	Metabolic/endocrine/cardiovascular	
65, 66	cg44913012	75	GGAGTCATAGGC AAATGTTTAAT- T AATT/gap CTGC T-CA-TATGCAC- ATCTGAAAGC- ATGA	AATT (65)	gap (66)			SILENT-NONCODING	kinase	Human Gene SWISSPROT-ID Q01813 PHOSPHOFRUCTOKINASE, TYPE C (EC 2.7.1.11) (PHOSPHOFRUCTOKINASE 1) (PHOSPHOHEXOKINASE) (PHOSPHOFRUCTO-1-KINASE ISOZYME C) (6-PHOSPHOFRUCTOKINASE, PLATELET TYPE) - HOMO SAPIENS (HUMAN), 784 aa	6.0	4bp deletion, 8 of 60	10 (10p15.3)	Metabolic/endocrine/cardiovascular	
67, 68	cg44913012	142	ATGAGACACA- CTCC- ACAGACAGCACG CACTGG-[A/G]G- CTGGTGG- GGCAGATGGGCA CTCGCGGATTAG GT	A (67)	G (68)			SILENT-NONCODING	kinase	Human Gene SWISSPROT-ID Q01813 PHOSPHOFRUCTOKINASE, TYPE C (EC 2.7.1.11) (PHOSPHOFRUCTOKINASE 1) (PHOSPHOHEXOKINASE) (PHOSPHOFRUCTO-1-KINASE ISOZYME C) (6-PHOSPHOFRUCTOKINASE, PLATELET TYPE) - HOMO SAPIENS (HUMAN), 784 aa	6.0	27 of 75	10 (10p15.3)	Metabolic/endocrine/cardiovascular	

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69, 70	cg44913012	134	ATCTGAAGC-ATGAGACACA-CTCC-ACAGACAGCAC[G/A]CACTGG-AG-CTGGTGG-GGCAGATGGGCACTCGC	G (69)	A (70)			SILENT-NONCODING	kinase	Human Gene SWISSPROT-ID Q01813 610 PHOSPHOFRUCTOKINASE, TYPE C (EC 2 7 1 1) (PHOSPHOFRUCTOKINASE 1) (PHOSPHOHEXOKINASE) (PHOSPHOFRUCTO-1-KINASE ISOZYME C) (6-PHOSPHOFRUCTOKINASE, PLATELET TYPE) - HOMO SAPIENS (HUMAN), 784 aa	610	15 of 75	10 (10p15 3)	Metabolic/endocrine/cardiovascular	
71, 72	cg44913012	1530	CTGTCC-AG-CC-GA-TTT-CTTT-GATCT-GGCCCTT-GG-C-[A/G]AAGCC-G-T-CA-A-A-GCCAT-CA-TAGATGG-CG-AG-CAT-CCTG	G (71)	T (72)			SILENT-NONCODING	kinase	Human Gene SWISSPROT-ID Q01813 610 PHOSPHOFRUCTOKINASE, TYPE C (EC 2 7 1 1) (PHOSPHOFRUCTOKINASE 1) (PHOSPHOHEXOKINASE) (PHOSPHOFRUCTO-1-KINASE ISOZYME C) (6-PHOSPHOFRUCTOKINASE, PLATELET TYPE) - HOMO SAPIENS (HUMAN), 784 aa	610	36 of 200	10 (10p15 3)	Metabolic/endocrine/cardiovascular	
73, 74	cg44913012	1630	TGGCCGTCGGCAAT-GCCC-ACGCG-C-ACA-G-CTGAGC-G-T-ACGGCT[C]GCGT-T-CA-TCC-CA-GC-CGCGGGTGCCCCC-ACGTTGATGA	G (73)	T (74)			SILENT-NONCODING	kinase	Human Gene SWISSPROT-ID Q01813 610 PHOSPHOFRUCTOKINASE, TYPE C (EC 2 7 1 1) (PHOSPHOFRUCTOKINASE 1) (PHOSPHOHEXOKINASE) (PHOSPHOFRUCTO-1-KINASE ISOZYME C) (6-PHOSPHOFRUCTOKINASE, PLATELET TYPE) - HOMO SAPIENS (HUMAN), 784 aa	610	35 of 200	10 (10p15 3)	Metabolic/endocrine/cardiovascular	
75, 76	94238747	1638	TTCT-CCGGGCCCACTGGATGGTGA-GGGGT--CCCGGTGCCCAG-GT[C]GGGGGCGGC-AGGCTCCACTGGGCACITGCTGAGA-G-CITGCGGCTT-GAGCAGCCGCTGGTC	T (75)	C (76)			SILENT-NONCODING		CD98		41 of 50	16q24 3	Metabolic/endocrine/cardiovascular	Immunology

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77, 78	95072341	286	TTTATACAATAC AT-ACAAATTA- TCA-GG-AATG-C- AAAAAAAAAA[A/ gap]CATAAATAAT GCCCATTT-A- CA-GG-TG-A-C-A- TTTTAAA-C-AA- TG-AAAAA-C- ACCAACGG	A (77)	gap (78)			SILNET NON- CODING	Kinase	sgk		46 of 200	5	Metabolic/endocrine/cardiovascular	Renal Disease
79, 80	95308696	445	AAAGGTGTGGAT G-AAGCAACCAT- CATTGACA- TTCTA-CTA-AG- CGAAACAATGCA [C/T]AGCG- TCAACA- GATCAAAGCAGC ATAT-C-TCC	C (79)	T (80)	Gln	STOP	Termination		lipocortin 1	1.60E-07	2 of 150	9	Metabolic/endocrine/cardiovascular	
81, 82	cg43064060	805	AGAAACAA- ATGCCAG-TATTG- TC-GATTI- CACAAAGTCCIT TCTGTCGGGATG TCACACAAACG[ga p/GTAG]CGATTGG TTTTGGACGAA- TGCCAAAGATCTG AGAAAGCAAAAC TGAAAGCAGAAA	gap (81)	GTAG (82)			Frameshift	nucl_recpt	Human Gene SWISSPROT-ID Q07869 PEROXISOME PROLIFERATOR ACTIVATED RECEPTOR ALPHA (PPAR-ALPHA) - HOMO SAPIENS (HUMAN), 468 aa [pctls SPTREMBL- ID Q16241 PEROXISOME PROLIFERATOR ACTIVATED RECEPTOR ALPHA - HOMO SAPIENS (HUMAN), 468 aa (fragment)	4.10E-254	4bp insertion polymorphism 3 of 10	22	Metabolic/endocrine/cardiovascular	
83, 84	cg106711057	775	CGAT-GG-CT-T- GG-TCTT-A- AGGTGCCT-A-A- CCTCCTCT- GCAGCT[A]TT- CTCAAAC-T-CAG- CCTGAGA- CATCCT-GG-C-C- GACTT- GCAAGAACT- CCA	T (83)	A (84)	Lys	Asn	NON- CONSERVATIVE	Peptide hormone	Acyl CoA Binding Protein		2 of 130	6q13-15	Metabolic/endocrine/cardiovascular	CNS Disorders

TABLE 1

SEQ ID	Sequence Calling Assembly	Base pos. of SNP	Polymorphic Sequence	Base before	Base after	Amino Acid before	Amino Acid after	Type of Change	Protein classification of CuraGen gene	Name of protein identified following a BLASTX analysis of the CuraGen sequence	p value	Allele Freq. (pred.)	Map Location	Therapeutic Area #1	Therapeutic Area #2
85, 86	cg106711057	477	AAGGCATGTGTC-TC-AG-TTTAGG-ATAAACACATGGCACAGTAA-CC-AAATCCAG-TCTCT-CATATCCCCG[C/T]ATTTTTCITTAGCTCTCTACTTTGTTGATGTAAG	C (85)	T (86)	Tyr	Cys	NON-CONSERVATIVE	Peptide hormone	Acyl CoA Binding Protein		3 of 180	6q13-15	Metabolic/endocrine/cardiovascular	CNS Disorders
87, 88	cg108881866	170	TTCAGCT-GCACATGAATAGAACAGCAAT-G-AGAGCCAGTCAGAA-GG-ACTTTGAAAAATTCAAT[G/A]AATCAAGTGAACTC-TT-GAAAAA-GGAT-CCAGGAAA-CGAA-GTGA-AG-CTAAAACTCTACGCGCTATATAAGCAGGCC	G (87)	A (88)	Met	Ile	Conservative	Peptide hormone	DBI-related Protein		10 of 60	6p24 1-25 3	Metabolic/endocrine/cardiovascular	Oncology
89, 90	cg108881866	741	G-CTGCC-AG-C-AA-GG-ATG-A-CTCAAT-CATCACTG-TTTT-AAC-AGG-AA-A-TGGTGA-CT[A/G]TT-ACA-G-TA-G-T-GGGAA-TGA-T-CTG-A-CTAAC-T-T-C-AC-TG-ATA-TT-CC-CC-C-T-G-GT-GG-AG-T-AG-AG-GAG	A (89)	G (90)	Cys	Tyr	NON-CONSERVATIVE	Peptide hormone	DBI-related Protein		3 of 200	6p24 1-25 3	Metabolic/endocrine/cardiovascular	Oncology

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SEQ ID	Sequence Calling Assembly	Base pos. of SNP	Polymorphic Sequence	Base before	Base after	Amino Acid before	Amino Acid after	Type of Change	Protein classification of CuraGen gene	Name of protein identified following a BLASTX analysis of the CuraGen sequence	p value	Allele Freq. (pred.)	Map Location	Therapeutic Area #1	Therapeutic Area #2
91, 92	cg108881866	851	ATA-TT-CC-CC-C-T-G-GT-GG-AG-T-AG-AG-GAG-AAAG-CTA-AAAATA[A/G]TG-CC-GT-TTTA-C-TGAGGGAA-T-TT-G-T-GGGCTGTTTATAGATTTT	A (91)	G (92)	Asn	Ser	NON-CONSERVATIVE	Peptide hormone	DBI-related Protein		3 of 200	6p24 1-25 3	Metabolic/endocrine/cardiovascular	Oncology
93, 94	cg108881866	1309	C-AC-TTTT-C-AG-AAAGAAG-TCTGGA--CCAGGC-T-GAAGGCA-TTTGC-AAAGCTT--CCCCC-AAAT-G[C/T]CTTG-AG-AATTT-C-AAAAG-AGG-TAAT-CA-GG-AAAAG-AGAGAG-A-G-AAAACTACAC-GCT-GTT-AATG-C-TGA-AGAAATG-CAAT-G-T-CC-TTCAG	C (93)	T (94)	Val	Ala	Conservative	Peptide hormone	DBI-related Protein		8 of 50	6p24 1-25 3	Metabolic/endocrine/cardiovascular	Oncology

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SEQ ID	Sequence Calling Assembly	Base pos. of SNP	Polymorphic Sequence	Base before	Base after	Amino Acid before	Amino Acid after	Type of Change	Protein classification of CuraGen gene	Name of protein identified following a BLASTX analysis of the CuraGen sequence	p value	Allele Freq. (pred.)	Map Location	Therapeutic Area #1	Therapeutic Area #2
95, 96	cg108881866	1404	AATTT-C-AAAAG- AGG-TAAT-CA- GG-AAAAG- AGAGAG-A--G- AAAAAACTACAC- GCT-GTT-AAATG-C- TGA- AGAATG[T/G] AAT-G-T-CC- TTCAG-GG- AAGATGG-CTATC- AGAT-GAA- TGCACAAAT- GCTGGGGIG- AACTT-CTTAT- CCAGAAAA- TCAAA	T (95)	G (96)	Cys	Trp	NON- CONSERV ATIVE	Peptide hormone	DBI-related Protein		2 of 40	6p24 1-25 3	Metabolic/endocrine/cardiovascular	Oncology